

**A large deletion on CFA28 omitting *ACSL5* gene is
associated with intestinal lipid malabsorption in the
Australian Kelpie dog breed**

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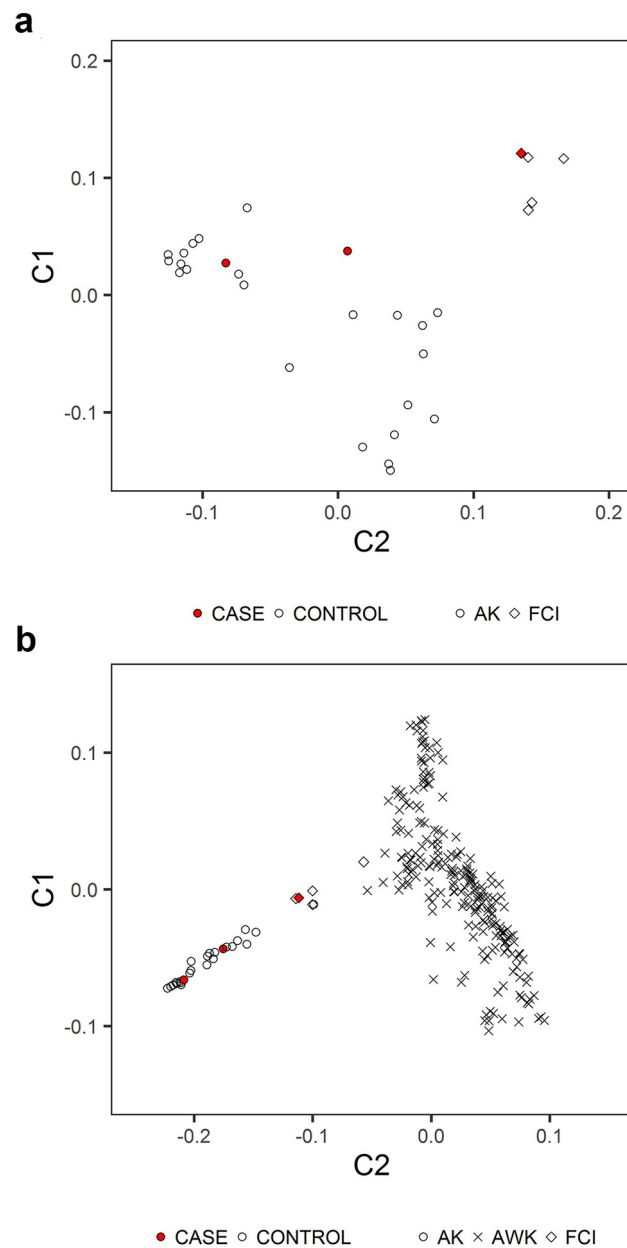
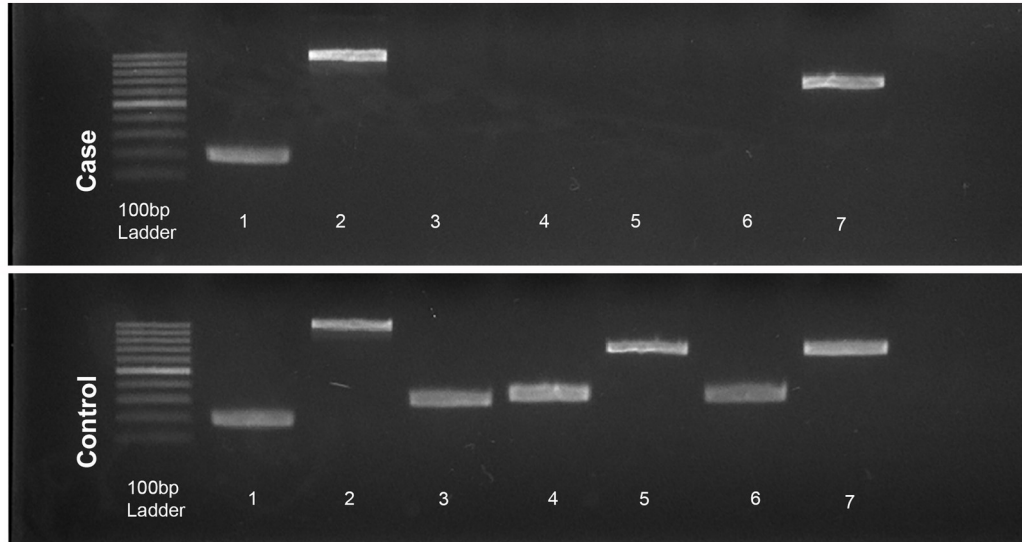


Figure S1. Multidimensional scaling plot of genotyped Kelpies.

MDS plot shows the distribution of case/control samples included in the GWAS. Kelpie registries differ in selective choices and geographical origins, samples have been labelled according to their respective registry. AK = Australian Kelpie; Australian population selected according to a conformation breed standard. AWK = Australian Working Kelpie; Australian Kelpie selected primarily for strong working ability. FCI = Federation Cytological International; International population allowing co-mingling of Australian populations. **(a)** dogs used in the primary association (n=30) **(b)** dogs included in the validation association (n=255).

a**b****Figure S2. Confirmation of deletion through PCR.**

(a) region on CFA28 (28:23370822-23493334), flanking a series of uncalled SNPs on canineHD BeadChip. Gene predictions are represented in red. Blocks are exons and untranslated regions, connected by introns (horizontal lines). Gene Symbols and arrows showing the direction of transcription are indicated in purple. 7 primer pairs were designed to amplify the edges of the suspected deletion. The regions amplified by the primers are coloured in blue. **(b)** PCR results. Lane numbers correspond to primer pairs illustrated in supplementary figure 1a, primer details outlines in Supplementary table 1. The top and bottom results come from a case and control respectively. In the control sample DNA amplification was consistently seen with all primer pairs, while it was only possible with primer pairs 1, 2 and 7 in cases. These results were indicative of a deletion between 101.6 kb and 105.2 kb in affected Kelpies. Gel image has been cropped to improve clarity and conciseness. Full gel image can be seen in the supplementary figure 3b.

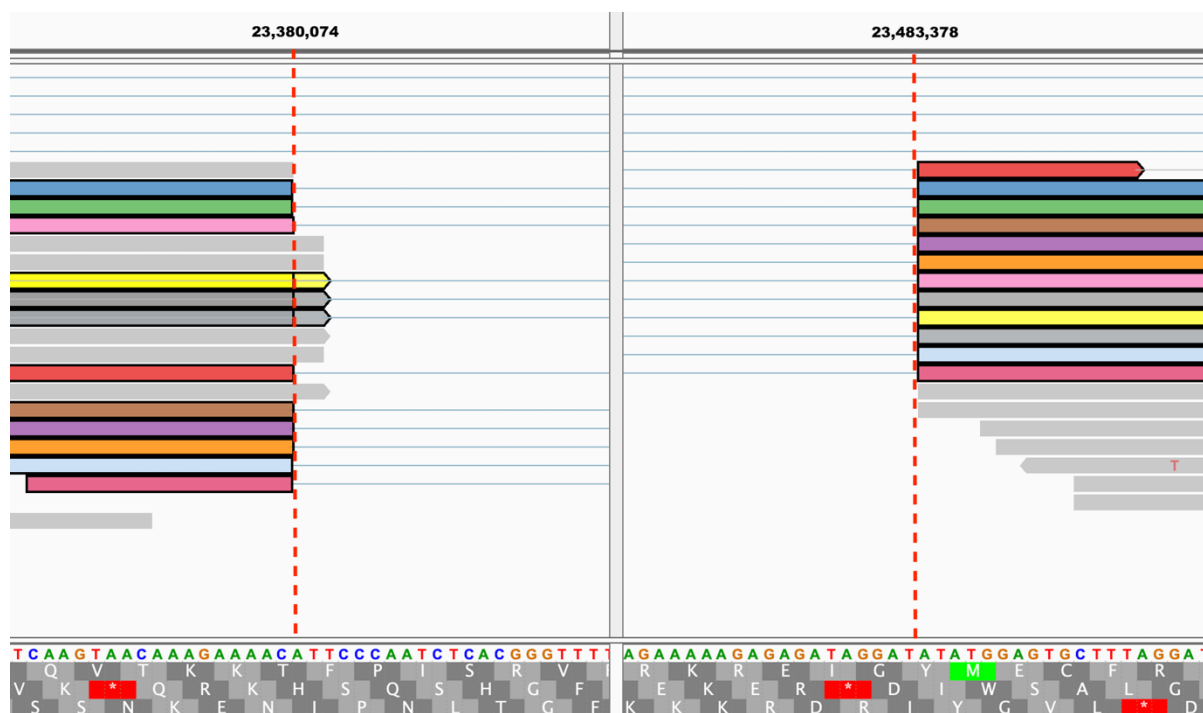


Figure S3. DNA reads from an affect Australian Kelpie mapped to CanFam3.1 captured a 103.3 kb deletion (NC_006610.3CFA28:g.23,380,074_23,483,377del). Positions are marked by dashed red lines. Paired reads flanking the gap are coloured the same. Visualised in IGV.

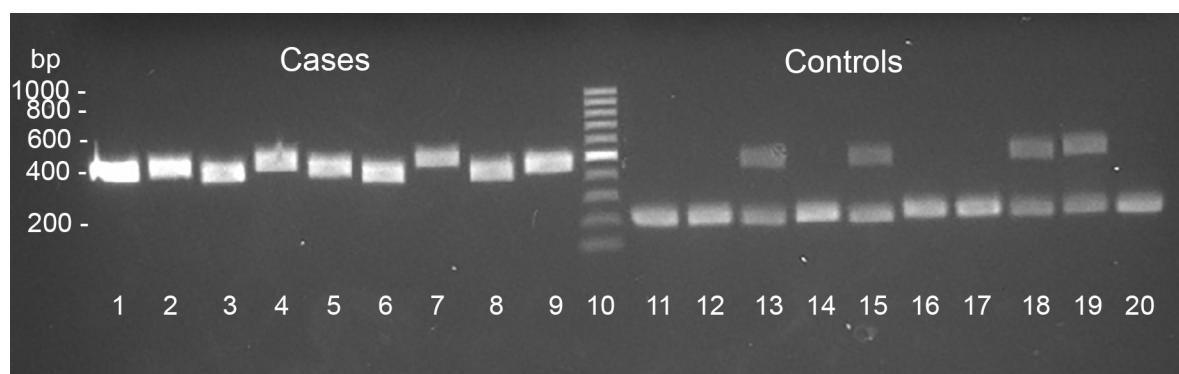
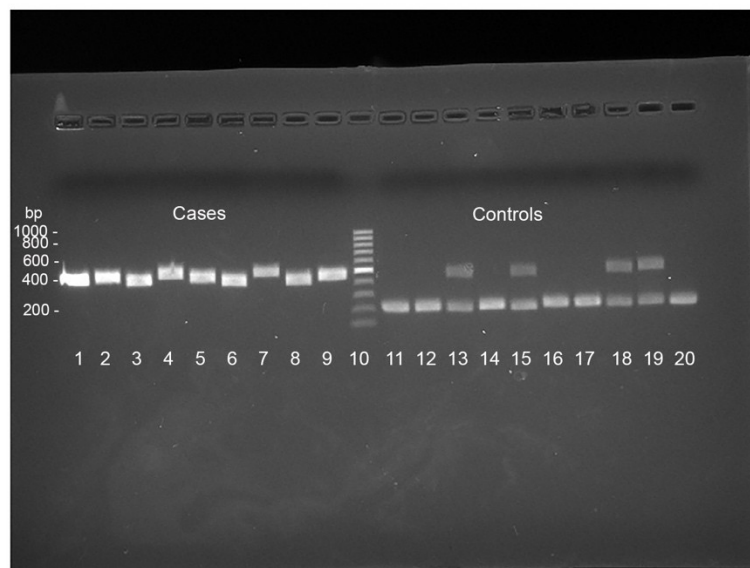


Figure S4. Multiplex-PCR diagnostic test results for 19 Kelpie samples.

Lanes 1-9 include case samples, each tested homozygous for the mutant allele. Lanes 11-20 include controls. All control samples with the exception of lanes 13, 15, 18 and 19 were homozygous wild-type while the others are carriers for the deletion. Lane 10 is the ladder (100 bp). All samples included were AK or international Kelpies. The trait has not been identified in AWK to date. Gel images have been cropped to improve the clarity and conciseness. Original Gel image can be seen in the supplementary figure 3a.

a



b

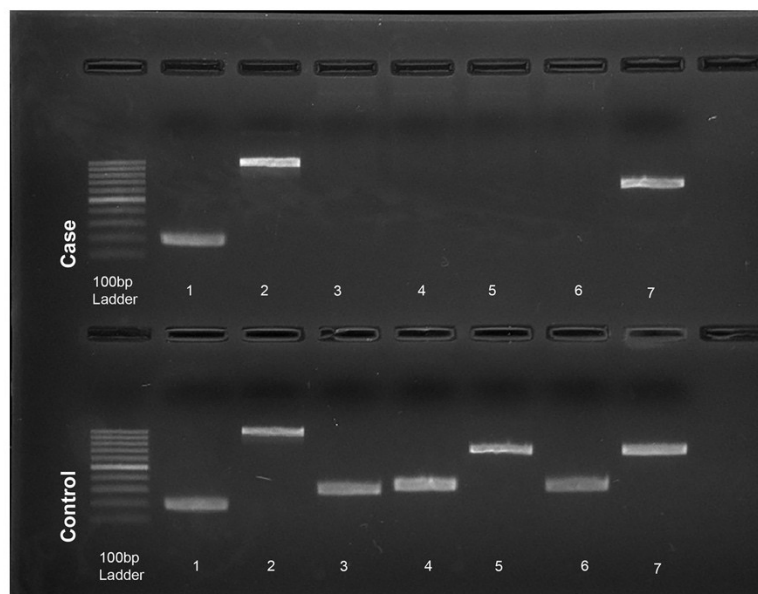


Figure S5. Full size, unedited PCR gel images.

(a) full scale image of PCR gel seen in figure 5. (b). full scale gel image from PCR gel included in supplementary figure S1b.

>AK_ZDHC6_prediction_Trinity_denovo_alignment

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Data S1. Consensus sequence of alternatively spliced ZDHC6 produced using a genome guided de-novo assembly with Trinity